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Clouston Syndrome: An Ectodermal Dysplasia Without Significant Dental Findings

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The ectodermal dysplasias are a heterogeneous group of conditions primarily affecting the hair, teeth, nails, and skin, and are classified according to the tissue(s) affected. The classification categories are: (1) abnormalities of hair, (2) dental defects, (3) abnormalities of nail morphology, and (4) dyshidrosis. Individuals are grouped according to defects present with findings from two or more categories required for diagnosis. As this classification method is based on phenotype, variable expression or small family size may well have an impact on diagnosis. We report a four-generation family with a hair-nail (1-3) dysplasia with nail morphology that is typical of Clouston syndrome. All affected relatives have thick, discolored, hyperconvex nails with onycholysis, varying degrees of hair involvement, and are hidrotic. They lack hyperkeratosis and multiple caries as originally described in Clouston syndrome. We propose that morphologic abnormalities of the teeth may not occur in the phenotype of Clouston syndrome and that it can be considered a hair-nail (1-3) dysplasia.

KEY WORDS: Clouston syndrome, ectodermal dysplasia, dystrophic nails, alopecia, keratoderma

INTRODUCTION

The ectodermal dysplasias are a heterogeneous group of conditions classified according to the tissue(s) affected, which are of ectodermal origin. Categories as designated by Freire-Maia and Pinheiro [1984] consist of: (1) abnormalities of hair, (2) dental defects (hypodontia, enamel hypoplasia, discoloration, abnormalities of size or shape) [Witkop et al., 1975], (3) abnormalities of nail morphology, and (4) dyshidrosis. Diagnosis requires the presence of defects in two or more of these categories. Abnormalities of other tissues of ectodermal origin may be present, as well as congenital abnormalities in other organ systems [Freire-Maia and Pinheiro, 1984]. We report a family with a hair-nail (1-3) dysplasia in 10 individuals in four generations (Fig. 1). Nail morphology and variable sparsity of hair in affected individuals was consistent with that described in Clouston syndrome. This family does not exhibit the multiple caries that were described in the original article by Clouston. Dental abnormalities have not been noted in other reported families with Clouston syndrome. Therefore, we propose that dental abnormalities, typical of the ectodermal dysplasias, are not a typical component of the Clouston syndrome.

CLINICAL REPORT

The proposita has dystrophic nails (Fig. 2) similar to those described and pictured in the report by Clouston [1929]. She also has sparse, slow growing hair and thin eyebrows (Fig. 3). There is normal distribution of hair on the remainder of the body, normal sweating, and normal teeth (Fig. 4), with no keratoderma. Her children were examined; her son is unaffected and her daughter has characteristic nails (Fig. 5) and slow growing hair with normal texture, amount, and distribution, normal teeth, and no keratoderma. By report, eight other relatives are affected, though they were not examined. The mother of the proposita is bald, and a half-brother and his daughter have very sparse scalp and body hair. There is an affected maternal uncle with normal scalp hair and no hair on the lower extremities. He has two sons and one daughter with similar phenotype. Characteristic dystrophic nails are present in all affected individuals in this family; however, there is wide variability in expression of hair abnormalities. No mildly affected individuals have had children who were more severely affected (Fig. 1). The presence of vertical transmission with an equal number of males and females affected and male-to-male transmission is consistent with autosomal dominant inheritance of a single mutant gene.

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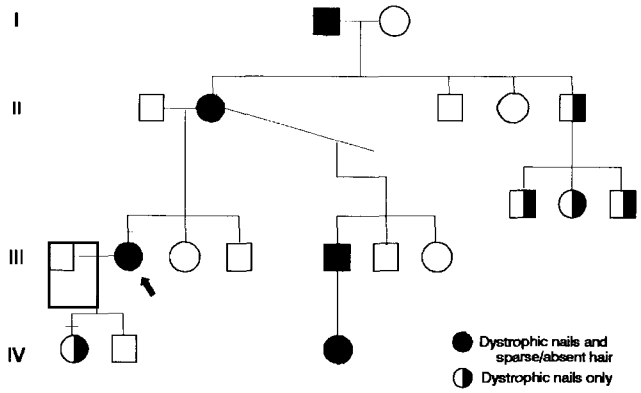

Fig. 1. Clouston syndrome. A family with hair-nail ectodermal dysplasia.

DISCUSSION

Jacobsen [1928] first reported on a family with brittle atrophic nails, sparse/absent or downy hair, "bad" teeth with marked caries, and rough scaly skin. All the affected relatives exhibited nail and teeth abnormalities and rough scaly skin. A large percentage also had absent or downy slow-growing hair over the entire body with the remaining affected individuals having patches of normal appearing hair. The family in this study was reviewed by Clouston [1929], who believed it to be part of a large French Canadian family he described. About 205 of the affected individuals had no abnormality of hair and the majority of those affected exhibited keratoderma. Although many had poor teeth with multiple caries, no morphologic dental abnormalities were reported. In his later review of ectodermal dysplasias,

Fig. 2. Dystrophic nails of proposita.

Fig. 4. The proposita with typical appearance of eyebrows and normal dentition.

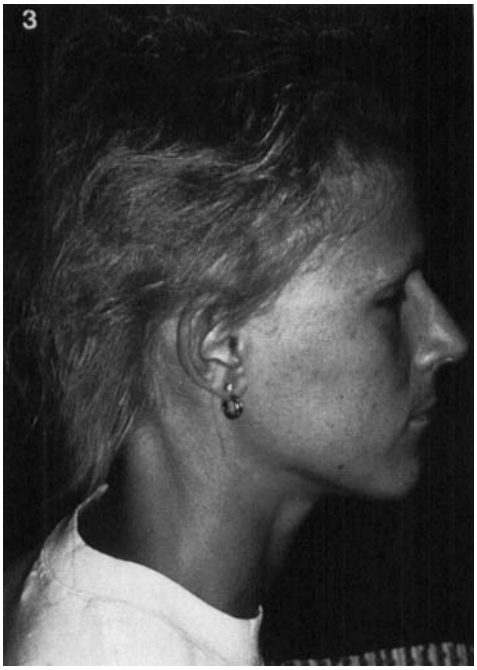

Fig. 3. The proposita with fine, sparse hair.

Fig. 5. Dystrophic nails of daughter of proposita.

Clouston [1939] reported that dysplastic nails were the most consistent feature of hidrotic ectodermal dysplasia, with keratoderma and abnormalities of the hair often present but to a variable degree. Abnormalities of the teeth were not included. Considering the dates of the early reports and the lack of dental findings described in more recent papers, the multiple caries reported in the initial evaluations may have been due to poor dental hygiene rather than to an underlying abnormality of dental morphology.

A number of other families with features of Clouston syndrome have been described in which dental abnormalities are notably absent. Wilkey and Stevenson [1945] reported on a family in Western Ontario thought to be of French extraction with 64 of 264 family members affected. They observed dystrophic nails in all affected relatives, abnormal hair in a majority, and keratoderma in some. All affected individuals had normal teeth. A seven-generation French Canadian family living in New Hampshire and Vermont showed dystrophic nails, alopecia, hyperkeratosis, normal facies, and normal sweating with no apparent dental abnormalities [Gagnon et al., 1989]. Rajagopalan and Tay [1977] reported on a Chinese family with similar findings in five generations. Dystrophic nails, abnormalities of hair, and keratoderma were reported. All affected individuals had normal sweating, normal facial appearance, and absence of dental abnormalities. In a family originally from Great Britain and reported on by Patel et al. [1991], all affected individuals had severely dysplastic nails, generalized alopecia, and keratoderma with no defects in size, shape, or number of teeth. Our family most closely resembles these pedigrees and that originally reported by Clouston.

Other forms of hair-nail dysplasia are distinct from the Clouston syndrome. A Brazilian family of German ancestry with a unique form of ectodermal dysplasia, in which the typical characteristics consisted of hypotrichosis, mildly dystrophic nails, and no dental defects, was reported by Pinheiro and Freire-Maia [1992]. That condition appears to differ from Clouston syndrome as it had hypotrichosis as the cardinal feature rather than dystrophic nails, which is the consistent feature in the Clouston syndrome. Finally, in contrast to the autosomal dominant disorders described above, Halal et al. [1991] described a family with similar clinical signs, e.g., dystrophic nails, sparse hair, normal teeth, and hidrosis, in which the condition was inherited in an autosomal recessive manner.

Although multiple caries have occasionally been reported in families with Clouston syndrome, morphologic dental abnormalities typically associated with ectodermal dysplasia have not been noted. Therefore, the Clouston syndrome fits the categorization of a hair-nail (1-3) dysplasia more accurately than its current classification as a hair-teeth-nail (1-2-3) dysplasia.

CONCLUSION

A distinctive nail morphology as described by Jacobsen [1928] and later by Clouston [1929], Wilkey and Stevenson [1945], Gagnon et al. [1989], Rajagopalan and Tay [1977], and Patel et al. [1991] is the single consistent finding in all affected individuals in the families reported. Hair abnormalities (fine, brittle, or sparse) show variability between family members and all affected individuals in all families described have normal teeth and sweating. The morphologic dental abnormalities which are components of the ectodermal dysplasias, i.e., natal teeth, missing and conical teeth, small or malformed teeth, delayed eruption, and enamel hypoplasia including pitting, were not reported in any of the families with Clouston syndrome. Therefore, we propose that morphologic abnormalities of the teeth need not be present to diagnose patients with the Clouston syndrome. It can be considered a hair-nail (1-3) dysplasia rather than a hair-teeth-nail (1-2-3) dysplasia as classified by Freire-Maia and Pinheiro [1984].

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